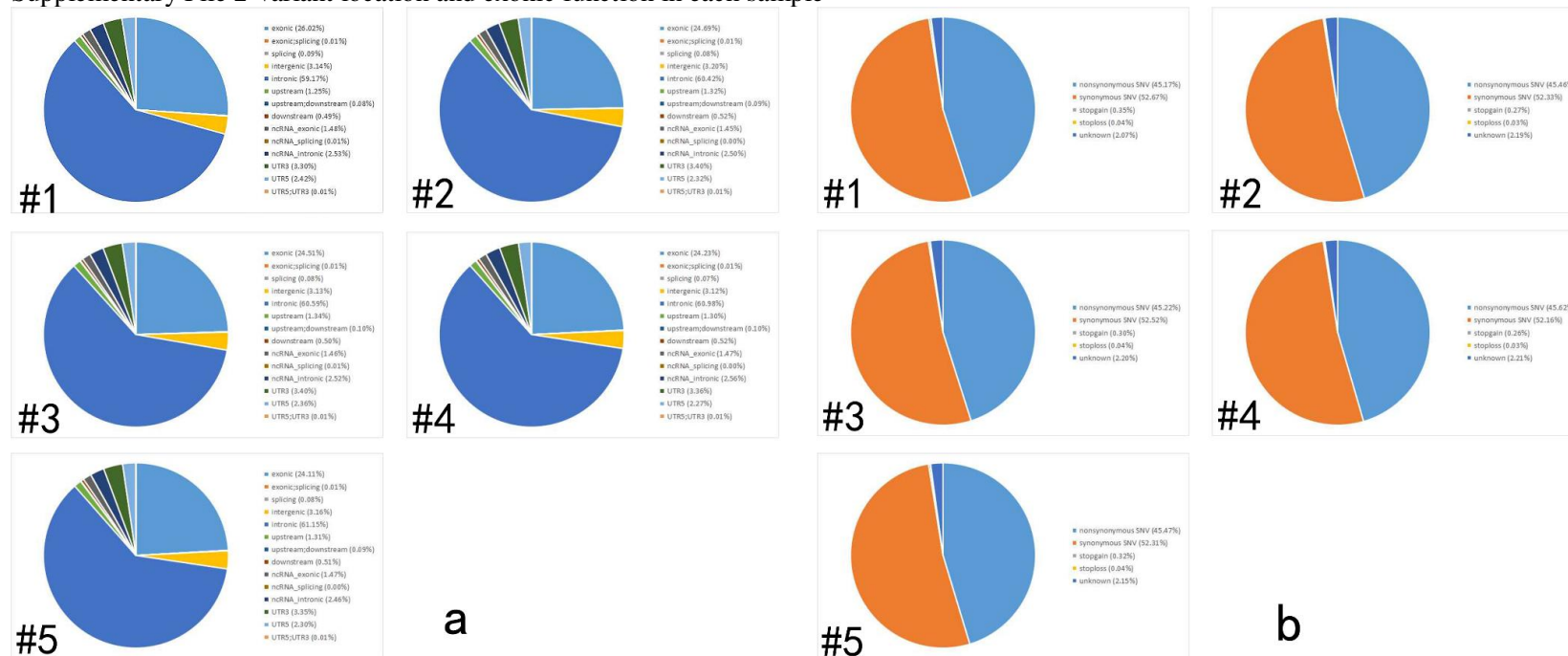


Supplementary File 2 Variant location and exonic function in each sample



Note: #1-5 was patient #1-#5

Note: Chr: Chromosome ID; Pos: mutation position in each chromosome; Ref: reference base; Alt: altered base; Alts: altered bases, if there is more than one altered base; P1–P5: patient 1–patient 5; 1000 G: allele frequencies in 1000 genomes project database.